

NEWBORN SCREENING INFORMATION MATRIX

Disorders Currently Screened in Washington

Disorder	Definition	Impact	Treatment	Benefits of Screen	Limitations of Screen	Comments
Congenital Adrenal Hyperplasia (CAH) (1 in 15,000 in WA)	Impaired production of cortisol and other adrenal hormones	Salt loss & shock may result in early sudden death, virilization & abnormal growth	Cortisol & salt-retaining hormone replacement	Prevention of salt crises & death, & reduction of virilization & abnormal growth	Can't reverse in-utero virilization of females; age of detection may not be early enough to prevent morbidity for all infants	Screening in WA since early 1984
Congenital Hypothyroidism (1 in 3,600 in WA)	Inadequate production of thyroid hormone	Mental retardation, growth failure	Thyroid hormone replacement	Prevention of mental & growth retardation	False positives increase with early specimen collection	Screening in WA since early 1977
Phenylketonuria (1 in 13,000 in WA)	Inability to break down phenylalanine, found in all foods containing protein	Mental retardation, seizures, & autistic-like disorders	Dietary restriction of phenylalanine using a special metabolic formula & specialty low-protein foods	Prevention of mental retardation and other neurological damage	Pregnancy risks for treated females	Screening in WA since early 1960's
Sickle Cell Disease (1 in 8,000 in WA)	Production of abnormal hemoglobin	Severe infections and possible death	Antibiotic prophylaxis to help prevent infections & parental education to recognize health crises	Decreased infections, morbidity & mortality	Transfusions can mask presence of disease	Screening in WA since 1991